NEUROMUSCULAR DISORDERS[1]

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The title "neuromuscular disorders" is perhaps not a very happy one but it permits discussion of all those conditions in which a muscle for some reason is not functioning as it should.

For a muscle to work properly, it must be intrinsically healthy, with healthy muscle fibres, normal in number, supplied with normally functioning motor end-plates. To these end-plates must run healthy peripheral nerve motor fibres, normal in number and capable of transmitting nervous impulses efficiently from the anterior horn cells in response to impulses travelling down the pyramidal tracts in the spinal cord from the motor cortex in the opposite cerebral hemisphere. The motor cortex itself is also played upon by a number of stimuli. Its responses depend on the patient's state of consciousness, on his emotional state and on the host of afferent impulses being received via the sensory network. Impulses from the extrapyramidal system, originating in the basal ganglia, play upon the pyramidal system and upon the muscle fibres with stretch receptors in the muscle spindles activating a controlling "seroo" circuit.

"Weakness" of a muscle may be due to a great many causes acting at any of these levels, and defining and localizing the cause may be a complex process.

LOCAL CAUSES IN THE MUSCLES

A muscle may not work properly because of pain. The pain may be the result of irritation, infection, or inflammation in adjacent connective tissue structures, be they joints or bones, or it may be referred from a visceral organ some distance from the muscle affected but supplied by the same peripheral nerve.

At times the muscle itself is inflamed, as in myositis. Trichinella spiralis may affect human muscle as well as pork and the patients present with acutely tender, painful muscles. In dermatomyositis, a diffuse disorder of collagen tissue, the overlying skin is reddened and inflamed, and the muscle is invaded with inflammatory cells. In polymyositis, an uncommon disorder, the muscle may be packed with inflammatory cells, and acutely tender to touch. Wasting and weakness may progress with alarming speed, and unless a remission can be obtained with steroid therapy progressive weakness may lead to complete paralysis and death from respiratory insufficiency. Both polymyositis and dermatomyositis, though rare, are very important to recognize, for with steroids a complete remission may be achieved.

The so-called muscular dystrophies are much more common. These are primary disorders of muscle fibres, usually developing in early life and tending to progress steadily uninfluenced by any medical measures. There is often a strong family history. They fall into four clinical groups.

The first group, formerly called Oppenheim's disease, is now termed benign congenital hypotonia, and describes the "floppy baby" which gradually recovers as it grows.

More common, unfortunately, is the group of pseudohypertrophic muscular dystrophies. The limb girdle muscles are severely affected, usually in boys of school age, and the calf muscles tend to be relatively well-preserved, presenting the appearance of pseudohypertrophy. The condition progresses over about seven years until the child is completely disabled. The patient assumes an attitude of lordosis when standing and develops a

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waddling gait. He rises from the ground by climbing up his knees in characteristic fashion.

Scapulohumeral and facioscapulohumeral muscular dystrophy affects a slightly older age group and affects both sexes. It is a much more benign disorder compatible with many years of survival. It has been said that the enigmatic smile of Mona Lisa was due to her suffering from this disorder. The electromyograph shows small very short duration (1 msec.) spike potentials on volition.

More striking than these disorders is dystrophia myotonica, the muscular dystrophy which is associated with so much muscle tone that when the patient shakes hands he is unable to release his grasp for a few seconds. These patients go bald prematurely, and may develop cataracts. They are handicapped both by their myotonia and by weakness, which often leads to foot-drop, and affects the muscles of the shoulder girdle, forearms and hands, as well as the sternomastoids and facial muscles. Walking calipers can be of great assistance. Life expectancy is good.

There is some evidence that the myotonia may be related to the relative concentrations of potassium within and without the muscle cell, and that it may be improved by altering this ratio by exchange resins, or by the use of cortisone or of procaine amide.

GENERALIZED DISORDERS CAUSING MUSCLE WEAKNESS

Familial periodic paralysis is a rare disorder characterized by the occurrence of sudden attacks of profound generalized muscle weakness. The attacks are associated with marked lowering of the serum potassium level and respond to elevation of the potassium level to normal. Similar attacks may occur when serum potassium levels are lowered by various diseases, such as diarrhoea or renal disorders, and by a low dietary intake, but in these there will not be a family history of affected relatives.

By liberation of acetyl choline at the motor end-plate, the termination of the nerve fibre at the muscle fibre, the nerve impulse is transmitted to the muscle fibre, which contracts. In myasthenia gravis there is a defect of acetyl choline production, or more probably, an excessive destruction of it by an enzyme, cholinesterase, and so transmission is faulty.

The patient, usually an adult, complains of excessive weakness, often of chewing or swallowing, of drooping of an eyelid, or weakness of elevation of the shoulders or extension of the fingers. The striking feature of this weakness is its variability from day to day, and from time to time in one day, the ready and fatiguability. A muscle may begin by acting strongly, as for example in combing and setting hair, but within a few minutes it may tire, so that the patient must drop the hands to rest, and after a short pause is able to resume again. Muscle wasting is not conspicuous in this disorder, in contrast to the severe weakness which is often present.

There is some evidence to suggest that the thymus gland is abnormally large in this disorder, and that it liberates an agent into the blood stream which blocks the motor end-plate. Certainly surgical removal of the thymus may cure the condition. Prostigmine given by mouth or injection will enable the motor end-plate to transmit efficiently, and many patients are maintained well for many years on this medication.

Thyrotoxicosis, or over-activity of the thyroid gland, is an uncommon cause of muscle wasting and weakness, but it is important because the results of medical treatment are so good, leading to full recovery of strength.

LESIONS AFFECTING THE NERVE SUPPLY

Lesions affecting the nerve supply to muscle are the commonest cause of muscle weakness and wasting. It is in this group that accurate diagnosis of the nature and site of the lesion is essential to enable successful treatment to be carried out, and it is in this group that preventive and remedial physiotherapy has so much to offer.

Disorders of Peripheral Nerves

Peripheral nerves can be affected in numerous ways and in numerous situations.

Bell's palsy is a common example. The facial nerve becomes acutely swollen as it travels down the narrow facial canal, presumably as a result of a virus infection. By swelling in so confined a space it becomes damaged and non-conducting so that weakness of the face develops on the affected side. The diminution of conducting power may be partial or complete, temporary or permanent, depending on the duration and severity of the nerve swelling. It is said that 80% of all cases of Bell's palsy will get useful recovery, but that if there is no improvement in eight weeks recovery is unlikely to occur. Various suggestions for improving the recovery rate have been made: opening the facial canal at the onset of the weakness, using anti-inflammatory agents such as cortisone, swing slings, massage, electrical stimulation and others, but the scientific evidence that any of these improves the results is still slender.

Other common nerve compressions are those of the ulnar nerve at the elbow and the median nerve beneath the flexor retinaculum in the carpal tunnel at the wrist. Transplantation of the ulnar nerve may be required for the first, whilst the second may respond to splinting, plastering, oestrogens, cortisone or diuretics, or if these fail, slitting of the flexor retinaculum may be required to provide relief.

The lateral popliteal nerve may be damaged as it passes laterally around the fibula, particularly by squatting, and foot-drop may result.

Trauma of various types may damage peripheral nerves or the brachial plexus.

Some disease processes may effect several or many peripheral nerves, and in these both motor and sensory abnormalities may occur. Toxic agents such as lead, arsenic, alcohol and organic phosphates (as in the appalling tragedy in Morocco two years ago) may produce generalized peripheral neuritis. Uncontrolled diabetes mellitus usually causes a neuritis affecting the sensory rather than the motor elements. Vitamin B deficiency (beri-beri) produces a mixture of weakness and sensory loss.

The most striking form of peripheral neuritis, in which the disabilities are at their worst most severe, is the so-called acute infective polyneuritis. No infective agent has been grown as yet, and it may be that the disease is a hypersensitivity reaction or allergic process. Often, muscle weakness is of rapid onset, and profound, affecting speech, swallowing, facial muscles, intercostals and diaphragm, as well as trunk and limb musculature. The patient is for a time, sometimes for weeks, at death's door, yet if kept alive in a respirator with good nursing he will make a full and complete recovery with good physiotherapy supervision.

Neurological Lesions at Other Level

Wasting and weakness of muscles may occur as a result of irritation or compression of nerve roots within the spinal canal, or as a result of diseased intervertebral discs in the cervical or lumbosacral regions, or less commonly, in the thoracic spine. Nerves may also be damaged by inflammatory thickening of the arachnoid, bony distortion or destruction at various sites, and by spinal cord tumours. In these conditions pain will usually be present, radiating along the course of the root, and sensory signs often accompany motor ones.

Another disorder in which muscle wasting accompanies muscle weakness, but in which there is no sensory loss, is that due to destruction of the anterior horn cells within the spinal cord. Acute destruction of anterior horn cells occurs in poliomyelitis, where the polio virus appears to pick out groups of anterior horn cells in a patchy way, some muscles being affected and others spared. There is no need to point out the immense value of physiotherapy in the rehabilitation of patients with this condition.

Less active anterior horn cell destruction occurs in motor neurone disease. In this

condition the shoulder girdle muscles and interossei are affected early, with rapid wasting and with abnormal visible twitches of muscle fibres known as fasciculation. A slow downhill course occurs over two to seven years. The lower limbs often show signs of a pyramidal lesion with a spastic weakness and increased tendon reflexes, and at times the swallowing mechanism is also affected. Medical management and physiotherapy have unfortunately no curative place here, but as means of maintaining morale in a chronic illness they are invaluable.

CONCLUSION AND SUMMARY

Muscular weakness may be the result of lesions affecting the upper or lower motor neurones and their pathways at a variety of levels. Localized lesions may have a local cause, such as pressure or vascular disease, or they may be the predominant manifestations of generalized disorders, either metabolic, infective or toxic. In addition, a group of conditions producing weakness affect primarily the muscles themselves: many of these are congenital, and in others the aetiology is largely obscure. The clinical features of a variety of disorders directly or indirectly affecting muscle power and efficiency are briefly described: an understanding of these features and of the essential pathophysiology is necessary to appreciate the role and objectives of physiotherapy in each of these conditions.